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An unusual occurrence of isolated thoracoschisis

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ABSTRACT

Thoracoschisis is a rare congenital malformation that is frequently associated with limb and abdominal wall defects. Here we present only the third case of isolated thoracoschisis with no associated limb abnormalities. A female infant was born at 38 weeks via C section and was found to have a chest wall defect measuring 2 × 2 cm superior to a low-set left nipple. Reduction of eviscerated abdominal contents including liver, stomach, and transverse colon with closure of overlying skin was successfully performed on first day of life. The unique findings in the current case could ultimately shed light on the pathophysiology of thoracoschisis and its embryological origins.

Thoracoschisis is a rare congenital anomaly characterized by the herniation of intra-abdominal organs through a thoracic wall defect. It may present itself as an isolated anomaly, or more commonly, in association with the limb-body wall complex (LBWC) [1–3]. The etiology of isolated thoracoschisis remains ill-defined, though the pattern of these defects does appear to provide some insight into the timing of the causative disruptive event in utero. There are only 2 previous cases of isolated thoracoschisis, with one survivor [5,6]. We present the first female case, and only the second survivor.

1. Case report

A 19-year-old mother underwent routine ultrasound at 23-week gestation during her first pregnancy. This demonstrated an otherwise normal appearing female infant with a skin covered mass consistent with abdominal contents protruding from the chest. Mid-pregnancy Alpha-fetoprotein (AFP) levels were noted to be normal. Additionally, the mother did not report any drug or medication use during pregnancy. The fetus was carried uneventfully to term. A female infant was subsequently born at 38 weeks via C-section weighing 2850 g. Upon birth she was found to have abdominal contents eviscerating through a chest wall defect measuring 2 × 2 cm superior to a low-set left nipple at the midclavicular line. There was no overlying amniotic sac or membrane present. Anterior portions of ribs 4–6 were congenitally absent. An orogastric tube was passed into the stomach, and this was visualized outside of the thorax on x-ray imaging. There were no limb abnormalities, and echocardiogram was normal (see Fig. 1).

Following 4 h of resuscitation in the neonatal intensive care unit, the baby was taken to the operating room for reduction of the evisceration. Initial attempts at reducing the abdominal contents through the chest wall defect were unsuccessful due to local adhesions. A separate left subcostal incision was made in order to reduce the abdominal contents through a presumed diaphragmatic hernia. From this incision, evaluation of the diaphragm was limited and no hernia was identified. Further investigation of the diaphragm was then undertaken via insertion of a 5 mm laparoscope through the surgically created subcostal incision, in the absence of pneumoperitoneum. This laparoscopic evaluation identified an intact diaphragm above the level of the chest wall defect. The

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Fig. 1. Pre-operative photo of abdominal contents eviscerating through 2 × 2 cm chest wall defect.

abdominal contents, containing portions of the liver, stomach, and transverse colon were then reduced via the subcostal incision. The skin overlying the chest wall defect was then closed primarily. Underlying soft tissue structures were not amenable to primary closure, and the decision was made not to close the chest wall defect with exogenous materials due to concerns surrounding the patient's expected rapid growth (see Fig. 2).

The patient had an unremarkable post-operative course. She was extubated on the first day post-operatively, began feeds 2 days later, weaned off oxygen and was otherwise ready for discharge on post-operative day 10. Ultimately she was discharged home with a customized external protective brace and plans for a staged chest wall reconstruction (see Fig. 3).

2. Discussion

Thoracoschisis is an exceptionally rare congenital malformation characterized by the herniation of abdominal contents though a thoracic wall defect. Previously, there have been seven case reports of thoracoschisis, with only two being isolated [1–3,5–8]. We describe the third case of isolated thoracoschisis, that is, an occurrence of thoracoschisis without any associated abnormality such as a diaphragmatic defect or limb-body wall complex. Additionally, our case represents the first female with isolated thoracoschisis, and the second surviving infant with this abnormality (see Table 1).

Our case is similar to the two previously reported cases of isolated thoracoschisis in that the chest wall defect was left-sided and less than 4 cm in diameter, the eviscerated abdominal contents had no covering amniotic sac, and the elevated left hemi-diaphragm to a level just above the thoracoschisis defect. These similarities would seem to provide some insight to the embryologic origins of

Fig. 2. Intra-operative photo taken after reduction of abdominal contents using a separate left subcostal incision.

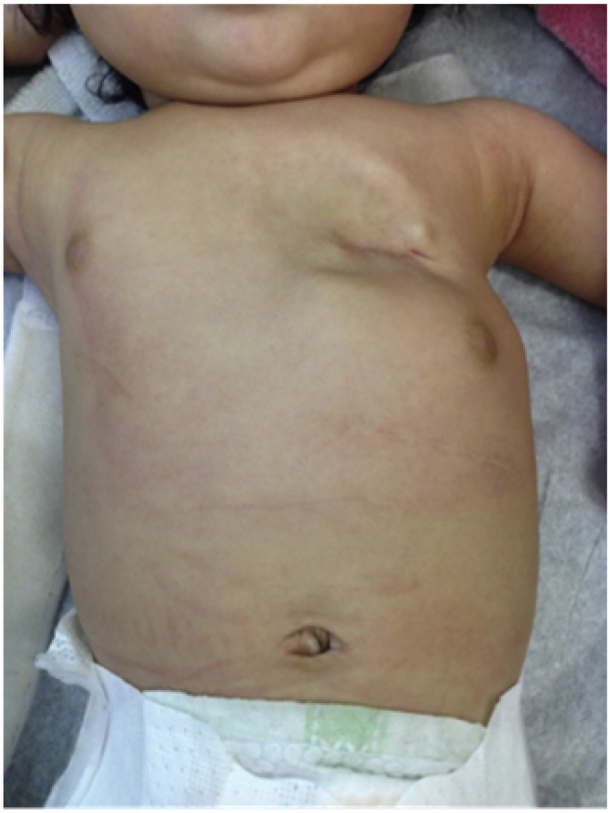

Fig. 3. Post-operative photo taken at 6 months of age. Plans made for a chest wall reconstruction before 1 year of life.

Table 1
Previous published cases of thoracoschisis.

Author	Year	Gender	Side	Diaphragmatic defect	Body wall defect	Limb defect	Outcome
Davies & Cywes [1]	1977	F	L	N	N	Y	Survived
Bamforth et al. [7]	1992	F	L	N	N	Y	Survived
Derbent & Balci [8]	2001	F	R	N	N	N	Intrauterine Death
Biri et al. [2]	2006	F	L	N	N	Y	Intrauterine Death
Isolated thoracoschisis cases							
Karaman et al. [5]	2011	M	L	N	N	N	Survived
Seleim et al. [6]	2014	M	L	N	N	N	Died DOL 2
Present Case	2015	F	L	N	N	N	Survived

this abnormality. It appears that the embryologic origin of thoracoschisis differs from that of gastroschisis. It is thought that the origin of gastroschisis may be linked to abnormalities in the lateral body wall folds responsible for closing the thoracic, abdominal, and pelvic portions of the ventral body wall medially. The body wall defects described in LBWC, and in the previously published cases of thoracoschisis, occurred in the lateral body and included an absence of skin, muscle, and peritoneum. The lateral body wall defect in LBWC suggests that the disruptive event occurred prior to the ventral fusion of the body wall [4].

Additionally, the intact elevated hemi-diaphragm seen in all three cases of isolated thoracoschisis may give us some insight to the timing of the formation the thoracic wall defect. It is possible that the development of the thoracic wall defect is secondary to a congenitally elevated hemi-diaphragm. In the absence of an elevated diaphragm, it would not be possible for herniation of intra-abdominal contents without diaphragmatic herniation.

The notable difference in our case, as compared to all of the previously reported cases of thoracoschisis, is the presence of segmental rib agenesis of ribs 4–6. The previous two cases of isolated thoracoschisis described only a widened intercostal space with increased separation of the associated ribs. It is feasible to hypothesize that the herniation of abdominal contents may have occurred at an earlier point in development in the current case, thus disrupting traditional rib formation.

It should also be noted that in our case normal AFP values were observed throughout pregnancy, further delineating this disease process from that of gastroschisis. Elevated AFP levels are usually seen in congenital abdominal wall defects, with more dramatic elevations being seen in gastroschisis as compared to omphalocele [9]. AFP values were not reported for the previous cases of isolated thoracoschisis, but have been found to be elevated in cases of thoracoschisis with associated diaphragmatic hernia [2]. These inconsistent elevations in AFP levels could be a direct result of the relatively lower amounts of the gastrointestinal tract exposed to the amniotic fluid during pregnancy in thoracoschisis as compared to gastroschisis.

3. Conclusion

Isolated thoracoschisis is a rare condition with only 2 prior reports and one previous survivor. The current case presents with unique findings which could ultimately shed light on the pathophysiology of this condition as well as its relations to traditional gastroschisis. Pre-natal findings of a chest wall mass or herniation of abdominal contents superior and lateral to the umbilicus should raise concern for thoracoschisis regardless of AFP levels.

Conflicts of interest

The authors declare that they have no conflicts of interest.

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